

AMENDMENTS TO THE CLAIMS

1. (Currently Amended) A method of diagnosing myelinopathy in an individual, said myelinopathy resulting from a periaxin (PRX) alteration in the individual, comprising the steps step of:

~~obtaining~~ assaying a sample ~~containing nucleic acid~~ from said individual for an alteration in a periaxin polynucleotide, said sample comprising periaxin polynucleotide genomic DNA having two PRX alleles, wherein said periaxin polynucleotide comprises SEQ ID NO:76 and wherein the presence of the alteration in both PRX alleles diagnoses said individual as having myelinopathy;

~~assaying said sample for an alteration in a periaxin polynucleotide, wherein; and said assaying step provides said diagnosis.~~

2. (Cancel)

3. (Cancel)

4. (Previously Presented) The method of claim 1, wherein said myelinopathy is selected from the group consisting of Charcot-Marie-Tooth (CMT) syndrome, hereditary neuropathy with liability to pressure palsies (HNPP), Dejerine-Sottas syndrome (DSN), congenital hypomyelinating neuropathy (CHN), and Roussy-Levy syndrome (RLS).

5. (Original) The method of claim 1, wherein said assaying step further comprises a polymerase chain reaction.

6. (Original) The method of claim 5, wherein primers for said polymerase chain reaction are selected from the group consisting of SEQ ID NO:3, SEQ ID NO:4, SEQ ID NO:5, SEQ ID NO:6, SEQ ID NO:7, SEQ ID NO:8, SEQ ID NO:9, SEQ ID NO:10, SEQ ID NO:11, SEQ ID NO:12, SEQ ID NO:13, SEQ ID NO:14, SEQ ID NO:15, SEQ ID NO:16, SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20, SEQ ID NO:21, SEQ ID NO:22, SEQ ID NO:23, SEQ ID NO:24, SEQ ID NO:25, and SEQ ID NO:26.

7. (Previously Presented) The method of claim 1, wherein said alteration is 3775G>A, 1216G>A, 4075-4077d, 1483G>C, 3394A>G, 3248C>G, 2763A>G, 2645C>T,

306C>T, 1491C>G, 2655T>C, 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.

8.-34. (Canceled)

35. (Currently Amended) A method of detecting the presence or absence of a mutation associated with a myelinopathy, said myelinopathy resulting from a periaxin mutation in the individual, the method comprising:

- a) isolating a test nucleic acid from a subject, said test nucleic acid comprising a periaxin polynucleotide of SEQ ID NO:76;
- b) comparing the test nucleic acid to a reference wild-type periaxin polynucleotide; and
- c) determining the differences between the test nucleic acid and the reference wild-type periaxin polynucleotide, wherein the differences are mutations in the periaxin polynucleotide of the subject, and wherein said detection of the presence or absence of the mutation is therein provided.

36. (Previously Presented) The method of claim 35, wherein said mutation is 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.

37. (Previously Presented) The method of claim 35, wherein said mutation encodes a defect of a periaxin polypeptide, wherein the defect is R953X, R368X, S929fsX957, R196X, V763fsX774, C715X, or R82fsX96.

38. (Cancel)

39. (Original) The method of claim 35, wherein said comparing step is by DHPLC, sequencing, hybridization, or a combination thereof.

40. (Previously Presented) The method of claim 35, wherein the myelinopathy is Charcot-Marie-Tooth (CMT) syndrome, hereditary neuropathy with liability to pressure palsies (HNPP), Dejerine-Sottas syndrome (DSN), congenital hypomyelinating neuropathy (CHN), or Roussy-Levy Syndrome (RLS).

42. (Previously Presented) The method of claim 35, wherein said mutation encodes a defect of a periaxin polypeptide, wherein the defect is E1259K, A406T, E1359del Δ , E495Q, R1132G, P1083R, I921M, A882V, T102T, P497P, or P885P.

43. (Currently Amended) A method of diagnosing myelinopathy in an individual comprising the ~~steps~~ step of:

~~obtaining~~ assaying a sample containing nucleic acid from said individual;

~~assaying said sample~~ for an alteration in a periaxin polynucleotide, said sample comprising periaxin polynucleotide genomic DNA having two PRX alleles, wherein said periaxin polynucleotide comprises SEQ ID NO:76 and wherein said alteration is associated with said myelinopathy, and wherein said myelinopathy comprises a prominent sensory neuropathy, wherein said assay provides said diagnosis.

44. (Cancel)

45. (Cancel)

46. (Previously Presented) The method of claim 43, wherein said assaying step further comprises a polymerase chain reaction.

47. (Previously Presented) The method of claim 46, wherein primers for said polymerase chain reaction are selected from the group consisting of SEQ ID NO:3, SEQ ID NO:4, SEQ ID NO:5, SEQ ID NO:6, SEQ ID NO:7, SEQ ID NO:8, SEQ ID NO:9, SEQ ID NO:10, SEQ ID NO:11, SEQ ID NO:12, SEQ ID NO:13, SEQ ID NO:14, SEQ ID NO:15, SEQ ID NO:16, SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20, SEQ ID NO:21, SEQ ID NO:22, SEQ ID NO:23, SEQ ID NO:24, SEQ ID NO:25, and SEQ ID NO:26.

48. (Previously Presented) The method of claim 43, wherein said alteration is 3775G>A, 1216G>A, 4075-4077d, 1483G>C, 3394A>G, 3248C>G, 2763A>G, 2645C>T, 306C>T, 1491C>G,

2655T>C, 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T₂ or 247ΔC.

49. (Currently Amended) A method of detecting a polymorphism or mutation in a periaxin polynucleotide of an individual, comprising the ~~steps~~ step of:

~~obtaining~~ assaying a periaxin polynucleotide comprising SEQ ID NO:76 for the polymorphism or mutation ~~a sample comprising said periaxin polynucleotide from~~ from a sample from said individual;

~~assaying said periaxin polynucleotide for the polymorphism or mutation.~~

50. (Cancel)

51. (Previously Presented) The method of claim 1, wherein said myelinopathy is Dejerine-Sottas syndrome.

52. (Previously Presented) The method of claim 1, wherein said individual is suspected of having the myelinopathy.

53. (Previously Presented) The method of claim 1, wherein the alteration comprises a homozygous periaxin mutation.

54. (Previously Presented) The method of claim 1, wherein the alteration comprises a compound heterozygous periaxin mutation.

55. (Previously Presented) The method of claim 43, wherein the alteration comprises a homozygous periaxin mutation.

56. (Previously Presented) The method of claim 43, wherein the alteration comprises a compound heterozygous periaxin mutation.

57. (Currently Amended) A method of identifying an individual suspected of having myelinopathy or being a carrier of myelinopathy, comprising the ~~steps~~ step of:

~~obtaining from said individual a sample comprising nucleic acid; and~~

assaying ~~said a sample from said individual, wherein said sample comprises nucleic acid,~~
for an alteration in a periaxin polynucleotide, ~~wherein said polynucleotide comprises SEQ~~
ID NO:76 and wherein the presence of the alteration identifies said individual as having
periaxin-associated myelinopathy or being a carrier of periaxin-associated myelinopathy.

58. (Previously Presented) The method of claim 57, wherein said myelinopathy
comprises a prominent sensory neuropathy.

59. (Previously Presented) The method of claim 57, wherein the alteration comprises
a homozygous periaxin mutation.

60. (Previously Presented) The method of claim 57, wherein the alteration comprises
a compound heterozygous periaxin mutation.

61. (Currently Amended) A method of identifying an individual suspected of having
myelinopathy or being a carrier of myelinopathy, comprising the ~~steps~~ step of:

~~obtaining from said individual a sample comprising genomic DNA having two PRX~~
~~alleles; and~~

assaying ~~said a sample from said individual, wherein the sample~~
~~comprises genomic DNA having two PRX alleles,~~ for an alteration in a
periaxin polynucleotide, ~~wherein said polynucleotide comprises SEQ~~
ID NO:76, wherein the presence of the alteration in the periaxin
polynucleotide is indicative of an alteration in at least one of the PRX
alleles, wherein the presence of the alteration in both PRX alleles
identifies said individual as having periaxin-associated myelinopathy
and wherein the presence of the alteration in one allele identifies said
individual as being a carrier of periaxin-associated myelinopathy.

62. (New) The method of claim 1, wherein the alteration in the periaxin polynucleotide
results in a loss-of-function for the corresponding periaxin polypeptide.

63. (New) The method of claim 35, wherein the alteration in the periaxin polynucleotide
results in a loss-of-function for the corresponding periaxin polypeptide.

64. (New) The method of claim 43, wherein the alteration in the periaxin polynucleotide results in a loss-of-function for the corresponding periaxin polypeptide.

65. (New) The method of claim 49, wherein the alteration in the periaxin polynucleotide results in a loss-of-function for the corresponding periaxin polypeptide.

66. (New) The method of claim 57, wherein the alteration in the periaxin polynucleotide results in a loss-of-function for the corresponding periaxin polypeptide.

67. (New) The method of claim 61, wherein the alteration in the periaxin polynucleotide results in a loss-of-function for the corresponding periaxin polypeptide.

68. (New) A method of identifying an individual with a myelinopathy, comprising the step of assaying a sample from an individual suspected of having myelinopathy for an alteration in a periaxin polynucleotide, wherein said polynucleotide comprises SEQ ID NO:76, wherein the presence of the alteration in the periaxin polynucleotide identifies said individual as having the myelinopathy.

69. (New) The method of claim 68, wherein the individual suspected of having myelinopathy is further defined as having one or more of a symptom selected from the group consisting of defective myelin, slowed motor nerve conduction velocity, muscle weakness, muscle atrophy, gait disturbance, onion bulb formations, palsy, areflexia, decreased sensitivity for touch, and decreased sensitivity for temperature.